

## Fahr's Syndrome- An Interesting Case Presentation

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### Abstract

### Case Report

Idiopathic basal ganglia calcification (IBGC), commonly referred to as Fahr's disease, is a rare neurological disorder characterized by the abnormal, symmetrical, and bilateral calcification of the basal ganglia and other brain regions. Patients typically present in their forties and fifties with various neurologic and/or psychiatric symptoms, including movement disorders, Parkinsonism, psychosis, and depression. The pathophysiology of this disease is not completely understood; however, several gene mutations have been identified in the pathogenesis of Fahr's disease. These mutations display an autosomal dominant inheritance pattern. Furthermore, the regional phenotypic expression of calcifications differs greatly from patient to patient, as do their clinical presentations. Here, we describe a patient who presented with psychiatric manifestations and imaging consistent with Fahr's disease.

**Keywords:** Fahr's syndrome, Bilateral intracranial calcification.

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## INTRODUCTION

Fahr syndrome is a rare neurodegenerative disorder determined by the presence of bilateral and symmetrical intracerebral calcifications of the basal ganglia. This condition is usually associated with phosphocalcic metabolism disorders, mainly secondary to hypoparathyroidism. It can be sporadic or familial. Chance discovery or during neuropsychic disorders, however, skin involvement can be revealing [1]. The diagnostic confirmation exam of choice is the brain CT scan. The current article reports a case of Fahr's syndrome due to hypoparathyroidism in cerebral palsy.

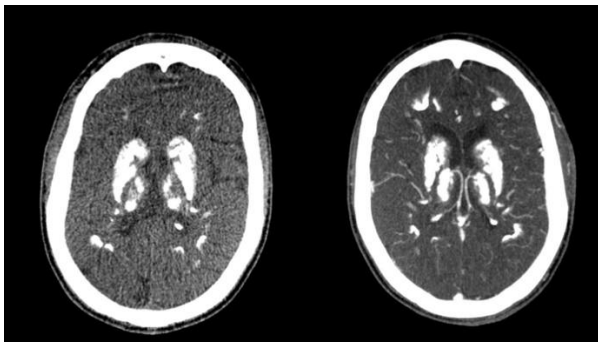
## CASE REPORT

A 30-year-old woman, with death story in siblings and follow-up for cerebral palsy since childhood, with mental retardation upon admission, the patient was confused and presented with a behavioral disorder, all evolving in a context of apyrexia and preservation of the general state. Physical examination revealed poor general condition, pallor, afebrile, anicteric and acyanotic. Her blood pressure was 125 / 79 mmHg and her heart rate was 78 beats per min. pulmonary auscultation revealed crackles in right hemithorax. There were spastic tetraparesis with muscle shortening and stiffness of respiratory and shoulder

girdle muscles. The laboratory tests showed hypocalcemia (total calcium of 3.9 mg/dL (normal level 8.4 - 10.3). White blood cell count and hemoglobin level were 15,500/mm<sup>3</sup>, and 9.5 g/dL, respectively. The levels of urea nitrogen and creatinine were normal (20 mg/dL and 0.7 mg/dL, respectively). The parathormone (PTH) level was 2 pg/mL (normal level 15 - 65 pg/mL). Chest radiograph showed alveolar infiltrate in the right lower lobe. CT of the brain revealed bilateral and symmetric, extensive, irregular, amorphous, calcifications involving the basal ganglia (caudate nucleus, globus pallidus, lentiform nucleus, and putamen), thalamus and dentate nucleus (**Fig-2**). Axial Noncontrast CT images of the brain in the level of posterior fossa show bilateral, dense and extensive calcifications involving the dentate nuclei and white matter of cerebellum with preserved cerebellar hemispheres. Note the "sun ray" calcification around the bilateral dentate nuclei (**Fig-1**). Axial Noncontrast CT images of the brain in the level of basal ganglia and lateral ventricles show bilateral, dense and extensive calcifications in the basal ganglia (caudate nucleus, globus pallidus, lentiform nucleus, and putamen). It is evident that the brain parenchyma does not show focal pathological changes. Calcifications are also seen in the choroid plexus of the ventricular system (**Fig-2**).



**Fig-1:** Noncontrast CT of the brain in the level of posterior fossa (A) and (B): Axial CT images show bilateral, dense and extensive calcifications involving the dentate nuclei and white matter of cerebellum with preserved cerebellar hemispheres in 30 years old women. Note the “sun ray” calcification around the bilateral dentate nuclei.



**Fig-2:** Contrast and noncontrast CT of the brain in the level of basal ganglia and lateral ventricles (A) and (B): Axial CT images clearly show bilateral, dense and extensive calcifications in the basal ganglia (caudate nucleus, globus pallidus, lentiform nucleus, and putamen) in 30 years old women. It is noticeable that the brain parenchyma does not show focal pathological changes. Calcifications are also seen in the choroid plexus of the ventricular system.



**Fig-3:** Coronal reformatted noncontrast CT scan images of the brain. (A) In the level of posterior fossa CT image clearly show bilateral, dense and extensive calcifications involving the dentate nuclei and white matter of cerebellum. (B) In the level of the frontal horns of the lateral ventricles, CT image clearly shows bilateral, dense and extensive calcifications in the basal ganglia (caudate nucleus, globus pallidus, lentiform nucleus, and putamen) in 30 years old women.

According to the clinical presentation, examination findings, and subsequent investigation exams, our case was diagnosed as Fahr’s syndrome due to a hypoparathyroidism. The evolution was favorable after the correction of the phosphocalcic disorders.

## DISCUSSION

Fahr syndrome is a rare entity whose causes are poorly understood. Its pathophysiology is not fully understood. In 1930, a German pathologist, Karl Theodor Fahr, described a case of a man with symmetrical calcifications of the basal ganglia and cerebral cortex [1, 2]. Some reports describe the inheritance of Fahr syndrome, mainly in an autosomal dominant way [2]. However, in the majority of patients, the syndrome does not have a genetic background. Bilateral basal ganglia calcifications can be observed in disorders of calcium and phosphorus metabolism, especially in hypoparathyroidism [4].

Fahr’s disease should be diagnosed based on clinical aspects, neuroimaging findings and the exclusion of other primary causes. It may occur in a sporadic or familial manner [4, 5]. Geschwind *et al.* [6], in a genetic study, described a dominant autosomal inheritance of the hereditary form of Fahr’s disease and suggested that the disease is caused by mutations in genes located on the long arm of chromosome [5].

The most common radiologic feature of Fahr’s disease is the presence of small bilateral intracranial calcifications which are usually restricted to the globus pallidus, but may also affect the putamen, caudate nucleus, thalamus, dentate nucleus and white matter of the cerebral hemispheres [5, 11-10]. Lester *et al.* [15] reported an atypical case of Fahr’s disease which developed stroke-like symptoms and progressed to extrapyramidal syndrome. Cranial CT demonstrated extensive bilateral and symmetric intracranial calcifications located at usual sites such as the basal ganglia and cerebellum and at unusual sites such as the temporal and frontal periventricular white matter and semioval centers. Intracranial calcifications can be detected incidentally in up to 0.3–1.2% of CT examinations of the brain. CT scanning is an easy test with maximum sensitivity and allows the easy diagnosis of Fahr’s disease [8]. Radiologists may detect bilateral abnormalities of the basal ganglia and thalamus in different acute and chronic clinical situations. The neuroimaging diagnosis is also influenced by detection of abnormalities involving other parts of the brain, especially the cerebral cortex, brainstem, and white matter. Important alternatives in the radiologic differential diagnosis for Fahr’s disease include hypoparathyroidism or pseudohypoparathyroidism (end-organ resistance to parathyroid hormone), which can be confirmed with measurements of serum calcium, phosphorus, and parathyroid hormone levels. Pseudohypoparathyroidism, in which there is no abnormality of calcium metabolism in asymptomatic

patients, is another possible diagnosis in patients with widespread cerebral calcification [23].

Some of the intracranial structures calcify and considered physiological with aging, are pineal gland 60%, habenular commissure 30%, choroids plexus 10%, dura mater 7%, petroclenoid, interclenoid ligaments 12%, pituitary gland and carotid arteries [24]. There are various causes of pathological calcifications in brain parenchyma, they may be infective, traumatic, metabolic and congenital conditions [21].

Intracranial calcifications may sometimes be observed on plain radiographs and frequently revealed by CT in normal older people. CT is the best modality to evaluate intracranial calcifications.

In conclusion, Fahr's syndrome is many times a treatable entity. Etiology is not directly correlated with image calcification pattern, except for some differences noticed in calcifications site in dystrophic senile ones. Topographic image studies are promising to predict neurological deficits. Their recognition by CT is easy, has maximum sensitivity and may be responsible by early treatment

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